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Xanthoma- Cutaneous Marker for Lipo-Protein Disorder

Dr. Anil Malleshappa[1], Dr. Smita Sonoli[2] , Dr.Vanishree Jabannavar[3], Dr. Manjunathswamy B.S[4]

1 Associate Professor, Dept of Biochemistry J N Medical College,KLES. Dr Prabhakar kore hospital & MRC Belgaum, INDIA
2 Associate Professor, Dept of Biochemistry J N MedicalCollege, Belgaum, INDIA
3 Assistant Professor, Dept of Biochemistry J N MedicalCollege, Belgaum, INDIA
4 Professor, Dept of skin & V.D J N MedicalCollege, KLES. Dr Prabhakar kore hospital & MRC Belgaum, INDIA
vanishreejabannavar@gmail.com, drbsswamy@yahoo.co.in

ABSTRACT:
Xanthomas are seen commonly in hyperlipedimias. Here we are reporting a rare case of type IIa hyperlipidemia diagnosed in 4 year old child who presented with multiple Xanthomas. This case report underlines the importance of recognizing xanthomas at the early stage so as to permit early diagnoses and treatment of the underlying hyperlipidemias and prevent the severe and inevitable coronary complication of lipid derangement especially in the children.

Key words: Xanthoma, Hyperlipidemia, Lipoprotein disorder.

INTRODUCTION

Many patients with lipoprotein disorders have increased risk of premature atherosclerosis and some develop Xanthomas and these acts as a cutaneous marker for the lipoprotein disorder.[1] Xanthomas are lesions due to accumulation of lipid-laden macrophages. Xanthomas are commonly caused by a disturbance of lipoprotein. They indicate a systemic alteration of cholesterol and / or triglyceride metabolism. When they do occur in children and adolescents a more severe form of hyperlipidemia should be suspected. Prompt diagnosis and treatment may help to prevent side effects such as early coronary artery disease.

CASE REPORT

A four year old female child presented with multiple asymptomatic yellow nodular swellings over gluteal cleft, upper posterior part of both thighs along the skin folds (fig.1), popliteal fossas, achilles tendon of both feet, few nodules coalesced to form plaques. Patient had no history of chest pain, headache, abdominal pain, jaundice, urinary complaints. There was no history of similar complaints in any of the family members, nor early death in family because of cardiac problem. History of consanguinity was present.

Lab studies: Routine investigations including Complete Heamogram, Liver function tests, renal function tests, Urine examination were all within normal limits.

Histological findings:
Histopathology Report showed: Epidermis is thinned and the dermis showed sheets of foamy macrophages which are admixed with a few touton type giant cells (fig.2 & 3) - Consistent with diagnosis of - Xanthoma**.**

Fasting lipid profile of patient revealed: (Table 1)

Table 1. Fasting lipid profile of patient.

Lipoprotein	Result	Ref. interval
Cholesterol	770 mg/dl	100-200
HDL Cholesterol	27 mg/dl	30-60
LDL Cholesterol	723 mg/dl	60-160
Triglycerides	99 mg/dl	30-200
VLDL Cholesterol	20 mg/dl	
Lipoprotein A	71.7 mg/dl	0-30

USG abdomen, Chest X-ray, ECG showed no abnormality. Patient was advised echocardiogram.

Fasting lipid profile of parents and three siblings: (Table 2)

Table 2. Fasting lipid profile of parents and three siblings.

Family members	Cholesterol (100-200)	HDL (30-70)	LDL (60-160)	Triglycerides (30-200)
Father	255	32	209	69
Mother	253	49	188	82
(sister)	220	28	176	78
(sister)	260	34	203	116
(brother)	183	29	137	83

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Figure 1. Yellow nodular swellings over gluteal cleft

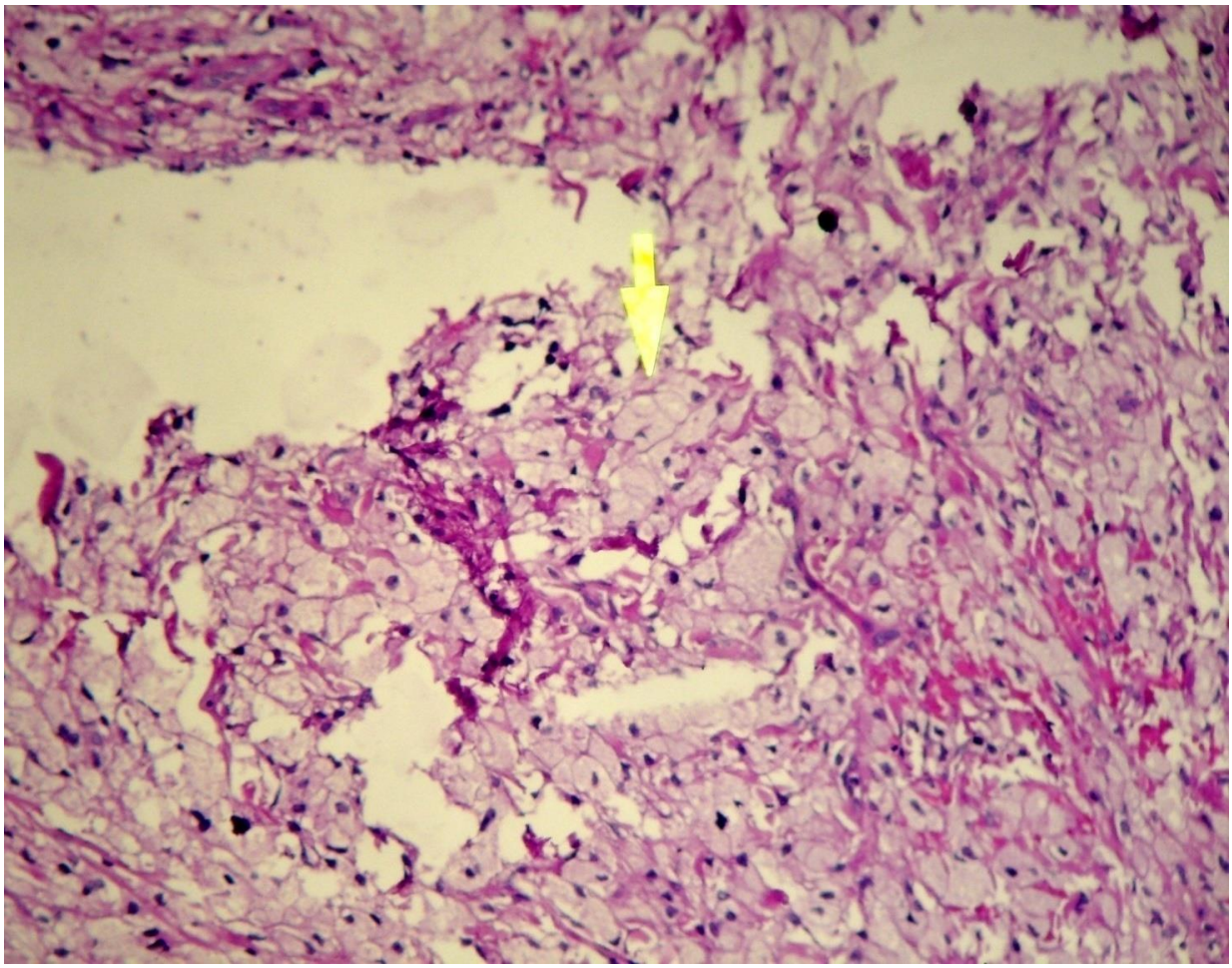

Figure 2. Showing foam cells [H& E, X400].

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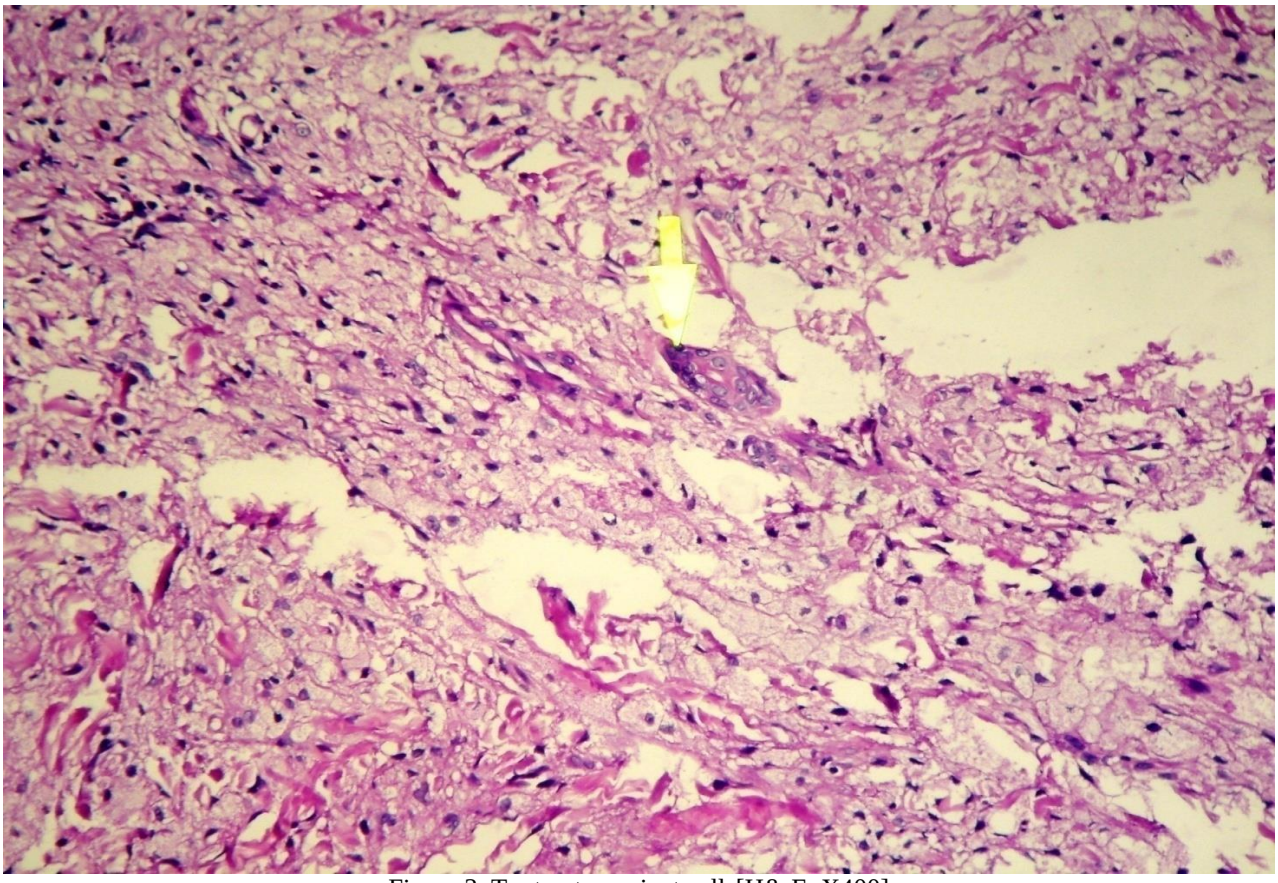

Figure 3. Touton type giant cells[H& E, X400]

Diagnosis of Familial hyperlipidemia in Homozygous form with type 2a pattern was made on the basis of increased levels of LDL in all of the family members.

DISCUSSION

Xanthoma may be seen either as a primary disorder caused by a inherited genetic condition of primary Hyperlipidemia or secondary hyperlipidemia due to various acquired systemic diseases like Diabetes mellitus, Biliary disease, Hypothyroidism, Nephrotic syndrome , etc.[2]An understanding of the pathogenesis of the underlying lipoprotein alterations provides a rational approach to therapy utilizing dietary manipulations and drugs.

Hyperlipidemia has been classified according to 6 phenotypes described by Frederickson's. The genetic and biochemical basis of these disorders has revealed a diverse group of diseases.Hyperlipidemia Type II a (Autosomal dominant):[3]Heterozygous form occurs in 1: 500, [3] homozygous form occurs in about 1:1,000,000. The primary defect is due to a reduction in LDL catabolism because of an abnormality in the LDL receptors. Heterozygotes express half the numbers of LDL receptors and homozygotes have between 0 -25 %.[4] Plasma cholesterol levels are severely elevated, but plasma triglycerides levels are typically normal. [4]Patients present with tendinous or tuberous xanthomas as well as xanthelasmas and may have severe atherosclerosis.Xanthomas may occur in persons of any age with no sex predilection. It usually presents as a cosmetic disorder (cutaneous xanthomas).[5] Their presence might suggest an underlying disorder of lipid metabolism (primary or secondary) for which they should be investigated.These cutaneous manifestations may precede a diagnosis of hyperlipidemia. Morbidity and mortality in these patients are related to atherosclerosis, pancreatitis. Xanthelasmas usually occur in people older than 50 yrs.

Treatment of xanthomas is, medical (drugs) for the underlying lipid disorders to prevent the complications of hyperlipidemia like cardiovascular and cerebrovascular accidents and surgical removal of deformity for cosmetic reasons. Medical treatment useful in such case is diet, lipid – lowering agents such as statins, fibrates, bile acid – binding resins, probucol, and nicotinic acid.[2]Eruptive xanthomas are said to resolve very early, tuberous xanthomas take about a month to resolve while tendinous xanthomas take years to resolve or may persist.

This case report underlines the importance of recognizing xanthomas at the early stage so as to permit early diagnoses and treatment of the underlying hyperlipidemias and prevent the severe and inevitable coronary complication of lipid derangement especially in the children.

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